

# Types of EB

## Epidermolysis Bullosa (EB)

EB-1

inherited EB  
EB hereditaria

Mechano-bullous → tendency to develop blisters following minor trauma / traction of skin

Cancer

Mutations in the genes encoding proteins of D<sub>ET</sub> → sub-epidermal or intraepidermal (15 structural proteins)

According to   
 { mutated gene   
 { mode of inheritance   
 { defective protein   
 { level of blister } 3 main types

Types of EB

EB

Innate bullous  
EBA

genetic  
EB hereditaria

EB

① Simplex (intraepidermal)

② Junctional

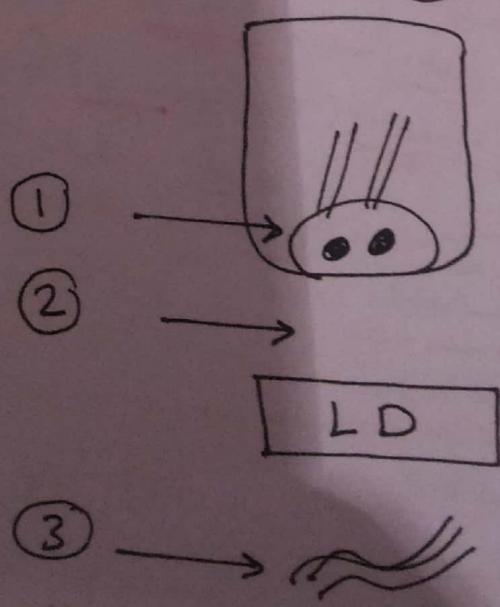
③ Dystrophic

Basal KCs in BMZ

LL

SLD

Tight As  
KS, 14



البربر

Update:

4 types

① simplex

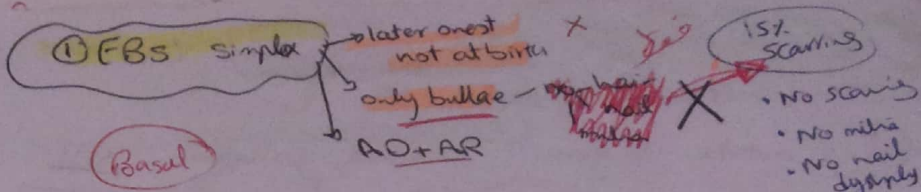
② JEB

③ DEB

④ Kindler's



- ① Suprabasal umbilical
- ② Basal → KS, K14



① AD

affected protein → KS, K14

② AR

affected protein → plectin → HD

① EBs with muscular dystrophy

② EBs with Pyloric atresia

Basal  
beachy

- ① localized EBs (Weber-Cockayne) (hyperhydrosis)
- ② Generalized severe (EBs herpiformis) (Dowling Meera) *grouped (polygenic)*
- ③ Generalized other (non Dowling Meera) (Kraeber)

④ EBs with mottled pigmentation (Rehmann hypopigmented macules on Trunk)

② JEB

AR

- with atrophic scar
- with Nail dystrophy
- enamel pits (Cobble Stone appearance)

Mentor defect in JEB

① generalized severe (Herlitz Type)

→ laminin 5 (332)

② generalized intermediate (non Herlitz type)

→ Lamin 5  
→ collagen XVII 17 (BPAG2)

③ JEB with pyloric atresia → α6β4 integrin

④ inverse type of JEB → intertriginous areas

Oris → cheilosis, c.p., R<sub>0</sub>

③ DER

- with atrophic scar ✓
- with Nail dystrophy ✓
- with milia ✓

Teeth → dental hypoplasia (Small teeth or irregular)

Collagen VII

① AD

Dominant DER

(DDER)

= peribulbar (skin only)  
= EB Pruriginosa only

② AR

Recessive DER (ROEB)

both collagen VII (SLO)

- ① generalized → 2 variants:
  - ① Cockayne Touraine
  - ② Passini Alboparmeloid
- ② localized:
  - ① Peribulbar
  - ② Peribulbar, nails only

① generalized severe (Hallopeau-Siemens)

② generalized intermediate (non Hallopeau-Siemens)

④ Kindler S → AR

protein defect

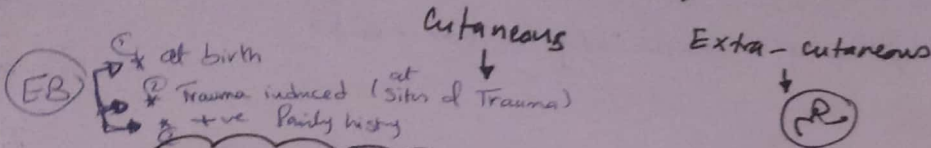
generalized

SLF



# Clinical picture

EB-3



## 1. Cutaneous findings

General characteristics (All types/subtypes):

- 1. Mechanically fragile skin → minimal trauma or movement of skin → blister formation

## 2. Erosions

## 3. Atrophic scarring (more in DEB > JEB > SEB)

EBs → NO x SCarring, NO nail, NO hair etc.

Question nail changes in EBD

- Dystrophic / absent nails
- Milia
- Scarring alopecia of scalp

## 2. Extracutaneous findings

any mucosal surface (like skin) → erosions → scarring

- Eye → Corneal blisters / ulcers / scarring / Ectropion
- oral cavity → microstomia / teeth hypoplasia
- Upper resp. tract → Tracheo-esophageal stenosis
- GIT → strictures / malnutrition / constipation
- Musculo-skeletal → muscle dystrophy (EBS-MD)

Extracutaneous complications more with EBD → more scarring

EBD > JEB > SEB

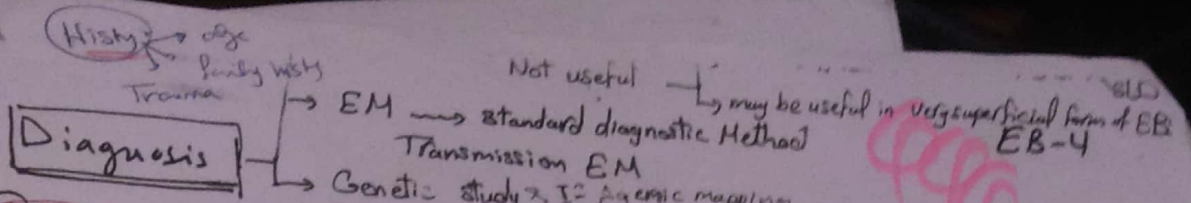
## 3. Complications

Skin malignancies especially in RDEB

(SCC - Melanoma)

- 1. well differentiated
- 2. frequently metastasizes
- 3. c.p. → indistinct borders (difficult to completely excise surgically)
- 4. no Resp. to Radio, chemo

Leading cause of death



① L/M (routine histopathology) : NOT useful

- × Can't differentiate level of cleft < <sup>IL</sup> <sub>SLD</sub>
- × may be useful in very superficial types of EBS

② E/M (standard diagnostic procedure)

→ Transmission EM (TEM)

③ Genetic studies / IF antigenic mapping?

To determine the defective gene / protein

• No Role to Histopathology

④ molecular diagnosis

**D.D.**

**EB  $\Delta$**

Mechanobullous disease

- chronic
- early onset
- +ve family history
- ve DIF

Trauma

• vesicles & bullae in neonates → HSV Bullous impetigo  
SSSS Bullous mastocytosis

• nail & teeth affection: → pachyonychia congenita

History

Bullae in neonates

① Trauma → sucking Blister

② Infect → HSV, SSSS, Bullous impetigo

③ Genetic → ① Bullous congenital ichthyiform erythroderma  
② Incontinentia pigmenti (vesicular stage)

④ Tumors → Bullous mastocytosis

⑤ Metabolic → acantholytic

⑥ autoimmune → Parapharyngeal pemphigus neonatal pemphigus acquired transplacentally

Blisters → erosion, excoriation

⑦ miliaria crystallina

⑧ Erythema multiforme



Liver • KBC • WBC •

Not useful - may be useful  
Standard diagnostic Method  
EM - mapping or protein

## Treatment of EB

EB-5

There's No specific TR for any type of EB

### General Measures

- Guard against
  - trauma
    - ① loose clothes
    - ② pads over bony prominence
  - infection
    - ③ anti-septic lotions
    - ④ antibiotics

### Skin care (dressings)

non-adhesive

### Systemic drugs

Topical phenytoin

HealSol

Spray

- phenytoin
- Collagenase
- Human collagen injection

↑ Collagen

Phibroblast activity → ↑ Fibrosis

### Others

- Gene therapy

↓ ID inject of collagen

⑦

~~Iso tretatinoin~~

low doses

it may ↑ mechanical fragility, blistering & skin

Chromoporphyrin

Suppress Collagenase

prevent scars

Hypertrophic scar

~~Colchicin~~

⊕ Collagenase

So used in Ref of Syst. sclerosis

cyclosporine and Thalidomide

Symptomatic relief of

EB Pruriginosa

Hemidesmosoma (EB)

Multilateral Gisting

- ①. AR
- ②. at birth
- ③. mutation

Kindlin-1 protein

Tremula liliacea

induced by

is a coral site

acid erosion

① bustens, evasions, progressive - deal with ship arap

② poikiloderm

Photoglossa

(4) ginseng

⑤ acquired

Stenosis

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defect

—

maculae hypermelanosis

Overion bacterium - a Cure

fecht-

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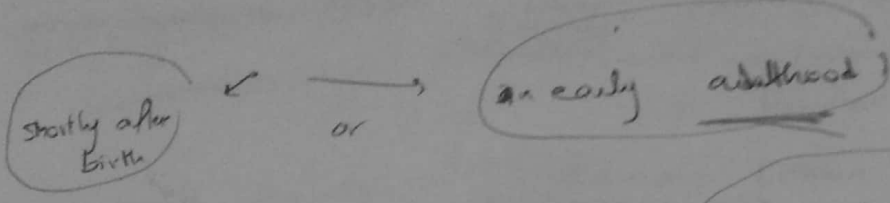
land

(localized form of EB)

EB pruriginosa

• rare subtype of dystrophic EB

- 1. Defect: mutation in gene coding collagen VII
- 2. C.P.: intensely pruritic hypertrophic plaques in linear configuration
  - lower extremities (peritibial skin) - most commonly
  - scarring + milium
  - tinea
- 3. Nail: Nail dystrophy is very common



- 3. DD: 1. prurigo nodularis, 2. hypertrophic L.P.
- 3. LSC, 4. lichen amyloidosis
- 3. Dermatitis, urticaria
- 4. Diag: 1. H.P. → hyperkeratosis, acanthosis, subepithelial blisters, perivascular lymphohistiocytosis

- 2. EM → level of split at SLD
- 3. DF → -ve

~~\* oral florid Papillomatosis~~  
~~\* florid type of endemic African Kaposi's Sarcoma~~

- 5. 1. Genod →
- 2. antibiotic →

- 2. cyclosporine, Papsone, Thalidomide, isotretinoin
- 4. UVB phototherapy → photo
- 5. Tacrolimus oint.

6. Course & complication  
1. Chronic, Refractory to Rx

2. complication → lymphoma  
SCC 222

Chronic